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AI Reports

Skin Health BETA

Genome: **Genome1**
Analysis: **AI Reports**
Data: **WGS**

Confidential Information

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Summary

Statistics

173 conditions screened
212 genes screened
7579 genetic variants screened

Key Findings

The genetic test results reveal a likely detection of a variant linked to Epidermolysis Bullosa Simplex (EBS), a genetic skin disorder known for causing fragile skin that blisters or tears easily. This condition is associated with a variant in the KRT14 gene, which plays a crucial role in maintaining the skin's structural integrity. Understanding this genetic predisposition empowers individuals to take proactive steps in managing their skin health. By being informed, you can collaborate with healthcare professionals to create a personalized care plan that mitigates symptoms and prevents potential complications. Resources such as patient advocacy organizations and support groups can provide valuable information and community support, enhancing your ability to manage the condition effectively. Current research is focused on improving treatment options and understanding the genetic mechanisms of EBS, offering hope for future advancements in care.

Next Steps

1. Consult a healthcare provider with expertise in dermatology to discuss the genetic sequencing results and potential implications for Epidermolysis Bullosa Simplex.
2. Schedule a genetic counseling session to understand the inheritance patterns, risks for family members, and implications of the genetic findings.
3. Consider additional diagnostic tests, such as immunofluorescence mapping or skin biopsy, to further investigate the specific subtype and severity of Epidermolysis Bullosa Simplex.
4. Explore available treatment and management strategies with a specialist to address symptoms and prevent complications associated with the condition.
5. Join a support group or connect with patient advocacy organizations for Epidermolysis Bullosa to gain access to resources and support networks.

Most Important Findings

(Red Category)

The genetic test results indicate a likely presence of a variant associated with Epidermolysis Bullosa Simplex (EBS), a condition characterized by fragile skin that can blister and tear easily. This condition is linked to a variant in the KRT14 gene, which affects the skin's structural integrity. While there is no cure for EBS, knowing about this genetic predisposition is empowering as it allows for proactive management of the condition. By understanding the genetic basis, individuals can take steps to protect their skin, manage symptoms, and prevent complications. This knowledge also provides an opportunity to work closely with healthcare professionals to develop a tailored care plan that supports skin health and overall well-being.

Epidermolysis Bullosa Simplex

Gene: KRT14
Variant Identifier: rs58645163 , RCV000487370
Your Data: TT
Risk Status: Likely Detected

Description: Epidermolysis bullosa simplex (EBS) is characterized by fragility of the skin (and mucosal epithelia in some cases) that results in non-scarring blisters and erosions caused by minor mechanical trauma. The current classification of epidermolysis bullosa (EB) includes two major types and 17 minor subtypes of EBS; all share the common feature of blistering above the dermal-epidermal junction at the ultrastructural level. The four most common subtypes of EBS are the focus of this GeneReview: EBS, localized (EBS-loc; previously known as Weber-Cockayne type). EBS, generalized intermediate (EBS-gen intermed; previously known as Koebner type). EBS-with mottled pigmentation (EBS-MP). EBS, generalized severe (EBS-gen sev; previously known as Dowling-Meara type). The phenotypes of these EBS types range from mild blistering of the hands and feet to more generalized blistering, which can be fatal. In EBS-loc, blisters are rarely present or minimal at birth and may occur on the knees and shins with crawling or on the feet at approximately age 18 months; some individuals manifest the disease in adolescence or early adulthood. Blisters are usually confined to the hands and feet, but can occur anywhere if trauma is significant. In EBS, gen intermed, blisters may be present at birth or develop within the first few months of life. Involvement is more widespread than in EBS-loc, but generally milder than in EBS-gen sev. In EBS-MP, skin fragility is evident at birth and clinically indistinguishable from EBS-gen sev; over time, progressive brown pigmentation interspersed with hypopigmented spots develops on the trunk and extremities, with the pigmentation disappearing in adult life. Focal palmar and plantar hyperkeratoses may occur. In EBS-gen sev, onset is usually at birth; severity varies greatly, both among and within families. Widespread and severe blistering and/or multiple grouped clumps of small blisters are typical and hemorrhagic blisters are common. Improvement occurs during mid- to late childhood. Progressive hyperkeratosis of the palms and soles begins in childhood and may be the major complaint of affected individuals in adult life. Nail dystrophy and milia are common. Both hyper- and hypopigmentation can occur. Mucosal involvement in EBS-gen sev may interfere with feeding, especially in neonates and infants. Blistering can be severe enough to result in neonatal or infant death.

Symptoms: Blisters on the skin, Erosions or raw areas on the skin, Thickened skin on palms and soles, Small white skin bumps (milia), Nail loss or deformity, Fragile skin that tears easily, Skin redness or inflammation, Pain and itching, Scarring and disfigurement, Difficulty

swallowing, Dental problems, Eye problems, Increased risk of skin cancer, Joint deformities, Growth retardation

Diagnostic Criteria: Clinical examination, Family history, Skin biopsy, Immunofluorescence mapping, Electron microscopy, Genetic testing, Blood tests for genetic markers, Prenatal genetic testing, Molecular analysis of keratin genes, Direct immunofluorescence

Treatment Options: Treatment for Epidermolysis Bullosa Simplex primarily focuses on symptom management and preventing complications. Key interventions include meticulous wound care to protect the skin, using non-adhesive bandages and moisturizing ointments to promote healing and reduce friction. Patients are often advised to make lifestyle adjustments, such as wearing soft, loose-fitting clothing and using padded shoes to minimize skin trauma. Pain management may involve the use of analgesics or other medications as prescribed by a healthcare provider. Additionally, maintaining a balanced diet and ensuring adequate nutrition can support skin health and overall well-being. Regular follow-up with healthcare professionals, including dermatologists and specialists in wound care, is essential for ongoing management and support.

Next Steps

1. Consult a healthcare provider with expertise in dermatology to discuss the genetic sequencing results and potential implications for Epidermolysis Bullosa Simplex.
2. Schedule a genetic counseling session to understand the inheritance patterns, risks for family members, and implications of the genetic findings.
3. Consider additional diagnostic tests, such as immunofluorescence mapping or skin biopsy, to further investigate the specific subtype and severity of Epidermolysis Bullosa Simplex.
4. Explore treatment options and management strategies with a specialist to address symptoms and prevent complications associated with the condition.
5. Join a support group or connect with patient advocacy organizations for Epidermolysis Bullosa to gain access to resources and support networks.

Additional Findings

(Orange Category)

No genetic conditions were found in this section.

Please note, this report is based on High-Confidence results only. If you were expecting to see conditions listed here, please check the 'Results' tab and make sure the 'Confidence' toggle is set to 'High-Only'.

Carrier Status For Inherited Conditions

(Yellow Category)

No genetic conditions were found in this section.

Please note, this report is based on High-Confidence results only. If you were expecting to see conditions listed here, please check the 'Results' tab and make sure the 'Confidence' toggle is set to 'High-Only'.

Medications That May Be Impacted By Your Genes

(Teal Category)

No genetic conditions were found in this section.

Please note, this report is based on High-Confidence results only. If you were expecting to see conditions listed here, please check the 'Results' tab and make sure the 'Confidence' toggle is set to 'High-Only'.

7570 – Total number of conditions screened.

To investigate more about these results, please visit **Next-Gen Disease Screen**.

No Increased Genetic Risk

(Green Category)

8 – Total number of conditions screened.

To investigate more about these results, please visit **Next-Gen Disease Screen**.

Unknown Genetic Risk

(Gray Category)

SAMPLE REPORT

List of Conditions Analyzed

Condition
Bloom Syndrome
Primary Ciliary Dyskinesia
Ciliary Dyskinesia
Cutis Laxa
Ehlers-Danlos Syndrome
Mucopolysaccharidosis, MPS-II
Partial Albinism
Oculocutaneous Albinism Type 3
Hereditary Leiomyomatosis And Renal Cell Cancer
Multiple Cutaneous Leiomyomas
Naxos Disease
Primary Ciliary Dyskinesia 5
Primary Ciliary Dyskinesia 7
Multiple Cutaneous And Mucosal Venous Malformations
Childhood Onset GLUT1 Deficiency Syndrome 2
Pontocerebellar Hypoplasia Type 3
Skin Fragility-Woolly Hair-Palmoplantar Keratoderma Syndrome
Woolly Hair-Skin Fragility Syndrome
Primary Ciliary Dyskinesia 2
Pontocerebellar Hypoplasia Type 2A
Skin/Hair/Eye Pigmentation, Variation In, 2
Acral Peeling Skin Syndrome
Stromme Syndrome
Epidermolysis Bullosa Simplex Due To Plakophilin Deficiency

Condition	Condition
Infantile Convulsions And Choreoathetosis	Ehlers-Danlos Syndrome, Musculocontractural Type
Primary Ciliary Dyskinesia 6	Combined Immunodeficiency With Skin Granulomas
Primary Ciliary Dyskinesia 12	Primary Ciliary Dyskinesia 11
Primary Ciliary Dyskinesia 10	Primary Ciliary Dyskinesia 9
Primary Ciliary Dyskinesia 7	Retinitis Pigmentosa, X-Linked, And Sinorespiratory Infections, With Or Without Deafness
Primary Ciliary Dyskinesia 13	Primary Ciliary Dyskinesia 14
Primary Ciliary Dyskinesia 15	Primary Ciliary Dyskinesia 16
Inflammatory Skin And Bowel Disease, Neonatal, 1	Developmental And Epileptic Encephalopathy, 1
Primary Ciliary Dyskinesia 20	Primary Ciliary Dyskinesia 17
Primary Ciliary Dyskinesia 18	Primary Ciliary Dyskinesia 19
Autosomal Dominant Nocturnal Frontal Lobe Epilepsy 5	Primary Ciliary Dyskinesia 21
Primary Ciliary Dyskinesia 22	Primary Ciliary Dyskinesia 23
Primary Ciliary Dyskinesia 24	Primary Ciliary Dyskinesia 26
Primary Ciliary Dyskinesia 27	Primary Ciliary Dyskinesia 28
Primary Ciliary Dyskinesia 29	Primary Ciliary Dyskinesia 30
Primary Ciliary Dyskinesia 33	Primary Ciliary Dyskinesia 32
Epidermolysis Bullosa Simplex 6, Generalized, With Scarring And Hair Loss	Paroxysmal Nonkinesigenic Dyskinesia 1
Multiple Benign Circumferential Skin Creases On Limbs 1	Galloway-Mowat Syndrome 1
Kartagener Syndrome	Episodic Kinesigenic Dyskinesia 1
Ciliary Dyskinesia, Primary, 40	Ciliary Dyskinesia, Primary, 43
Dyskinesia With Orofacial Involvement, Autosomal Dominant	Generalized Epilepsy-Paroxysmal Dyskinesia Syndrome
Cowden Syndrome 5	Cowden Syndrome 6

Condition	Condition
Cowden Syndrome 1	Bullous Ichthyosiform Erythroderma
Epidermolysis Bullosa Dystrophica	Epidermolysis Bullosa Simplex
Junctional Epidermolysis Bullosa	Recessive Dystrophic Epidermolysis Bullosa
Junctional Epidermolysis Bullosa Gravis Of Herlitz	Junctional Epidermolysis Bullosa, Non-Herlitz Type
Kindler Syndrome	Ichthyosis Bullosa Of Siemens
Generalized Dominant Dystrophic Epidermolysis Bullosa	Laryngo-Onycho-Cutaneous Syndrome
Epidermolytic Palmoplantar Keratoderma	Localized Epidermolytic Hyperkeratosis
Nephrotic Syndrome - Deafness - Pretibial Epidermolysis Bullosa Syndrome	Arrhythmogenic Cardiomyopathy With Woolly Hair And Keratoderma
Arrhythmogenic Cardiomyopathy with Wooly Hair and Keratoderma	Lethal Acantholytic Epidermolysis Bullosa
Crouzon Syndrome-Acanthosis Nigricans Syndrome	Hypertrophic Osteoarthropathy, Primary, Autosomal Recessive, 2
Isolated Focal Non-Epidermolytic Palmoplantar Keratoderma	Hypertrophic Osteoarthropathy, Primary, Autosomal
Junctional Epidermolysis Bullosa With Pyloric Atresia	Ellis-Van Creveld Syndrome
Medulloblastoma	Polyglandular Autoimmune Syndrome, Type 1
Hypohidrotic X-Linked Ectodermal Dysplasia	Hidrotic Ectodermal Dysplasia Syndrome
Johanson-Blizzard Syndrome	Jeune Thoracic Dystrophy
Brittle Cornea Syndrome 1	Hypoplastic Enamel-Onycholysis-Hypohidrosis Syndrome
Cranioectodermal Dysplasia 1	Odonto-Onycho-Dermal Dysplasia
Oculodentodigital Dysplasia	Cardio-Facio-Cutaneous Syndrome
Tooth Agenesis, Selective, 4	Oligodontia-Cancer Predisposition Syndrome
Ectodermal Dysplasia And Immunodeficiency 1	CHIME Syndrome
EEM Syndrome	Ectrodactyly, Ectodermal Dysplasia, And Cleft Lip-Palate Syndrome 3

Condition	Condition
Ectodermal Dysplasia And Immunodeficiency 2	CLOVES Syndrome
Cleft Lip/Palate-Ectodermal Dysplasia Syndrome	Cranioectodermal Dysplasia 2
Cranioectodermal Dysplasia 4	MEGF10–Related Myopathy
Ectodermal Dysplasia 10B, Hypohidrotic/Hair/Tooth Type, Autosomal Recessive	Ectodermal Dysplasia 10A, Hypohidrotic/Hair/Nail Type, Autosomal Dominant
Severe Growth Deficiency-Strabismus-Extensive Dermal Melanocytosis-Intellectual Disability Syndrome	Developmental Delay With Short Stature, Dysmorphic Facial Features, And Sparse Hair
Congenital Heart Defects And Ectodermal Dysplasia	Weill-Marchesani Syndrome 1
Linear Nevus Sebaceous Syndrome	Ectodermal Dysplasia 14, Hair/Tooth Type With Or Without Hypohidrosis
Ectodermal Dysplasia With Facial Dysmorphism And Acral, Ocular, And Brain Anomalies	Cardiofaciocutaneous Syndrome 1
Chédiak-Higashi Syndrome	Xeroderma Pigmentosum
Adrenoleukodystrophy	Familial Melanoma
Melanoma, Cutaneous Malignant, Susceptibility To, 2	Melanoma, Cutaneous Malignant, Susceptibility To, 3
Melanoma-Pancreatic Cancer Syndrome	Melanoma, Cutaneous Malignant, Susceptibility To, 5
Melanoma, Cutaneous Malignant, Susceptibility To, 8	Melanoma, Cutaneous Malignant, Susceptibility To, 10
Body Mass Index Quantitative Trait Locus 20	Oculocutaneous Albinism
Hermansky-Pudlak Syndrome	Tyrosinase-Positive Oculocutaneous Albinism
Tietz Syndrome	Hermansky-Pudlak Syndrome 2
Oculocutaneous Albinism Type 1B	Oculocutaneous Albinism Type 4
Griscelli Syndrome Type 2	Hermansky-Pudlak Syndrome 1
Tyrosinase-Negative Oculocutaneous Albinism	Tuberous Sclerosis Syndrome
Tuberous Sclerosis 1	Tuberous Sclerosis 2
Sjögren-Larsson Syndrome	Ichthyosis Vulgaris
Autosomal Recessive Congenital Ichthyosis 4B	Autosomal Recessive Congenital Ichthyosis 4A

Condition	Condition
DK1–Congenital Disorder Of Glycosylation	Autosomal Recessive Congenital Ichthyosis 11
CEDNIK Syndrome	Ichthyosis Prematurity Syndrome
Neutral Lipid Storage Myopathy	Autosomal Recessive Congenital Ichthyosis 5
Autosomal Recessive Congenital Ichthyosis 6	Weill-Marchesani 4 Syndrome, Recessive
Autosomal Recessive Congenital Ichthyosis 3	Autosomal Recessive Congenital Ichthyosis 10
Autosomal Recessive Congenital Ichthyosis 2	SRD5A3–Congenital Disorder Of Glycosylation
HELIX Syndrome	Ichthyosis, Congenital, Autosomal Recessive 13
Autosomal Recessive Congenital Ichthyosis 1	Wiskott-Aldrich Syndrome
Dermatitis, Atopic, 2	Generalized Pustular Psoriasis
Acrodermatitis Continua Suppurativa Of Hallopeau	

Glossary

KRT14 gene: A gene that provides instructions for making keratin 14, a protein essential for the structural framework of skin cells.

Keratin: A type of protein that is a key structural component of the skin, hair, and nails.

Genetic predisposition: An increased likelihood of developing a particular disease based on a person's genetic makeup.

Variant: A variation in the DNA sequence that may affect how genes function and can be associated with certain conditions.

Immunofluorescence mapping: A laboratory technique used to visualize the presence and location of specific proteins in tissue samples using fluorescent dyes.

Skin biopsy: A procedure in which a small sample of skin tissue is removed for examination under a microscope.

Electron microscopy: A technique that uses a beam of electrons to create detailed images of the ultrastructure of cells and tissues.

Genetic counseling: A process where individuals receive guidance and information about the genetic aspects of health conditions.

Patient advocacy organizations: Groups that provide support, information, and advocacy for individuals affected by specific health conditions.

Symptom management: Strategies and treatments aimed at reducing or controlling the symptoms of a condition.

References

Understanding Your Gene Inheritance Pattern

The Gene Inheritance Pattern explains how certain health conditions are passed down in families, focusing on a general pattern for each gene. While most variants in a gene are passed down in the same way, sometimes a specific variant might follow a different rule. For instance, even if a gene typically passes a condition in a dominant way (meaning only one copy from one parent can cause a condition), a particular variant within that gene might be recessive (meaning you need two copies, one from each parent, for the condition to appear). Therefore, it's important to consider that the way a specific variant is inherited could vary from the general pattern we describe for the entire gene.

References and Sources of Information

ClinVar (National Library of Medicine): **https://www.ncbi.nlm.nih.gov/clinvar/**
Genetic Home Reference: **https://ghr.nlm.nih.gov/**
National Institutes of Health: **https://www.nih.gov/**
ClinicalTrials.gov: **https://clinicaltrials.gov/**

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Remember that genetic information is one part of your overall health picture. It is essential to consider your family history, lifestyle, and other health factors in consultation with healthcare professionals.